

Table 2. Mutations in *VPS13A*

Exon	DNA Change ¹	Protein Change ¹
Intron 3	188-5T>G	Splice site mutation
4	237del	E80KfsX10
4	269T>A ²	I90K
Intron 6	495+1G>A	Splice site mutation
9	622C>T	R208X
Intron 11	883-1_892del	Splice site mutation
13	994del	A332LfsX9
13	1115del	K372SfsX1
13	1125_1128del	S375RfsX22
14	1187_1188del	F396fsX0
14	1208_1211del	Q403RfsX5
17	1549G>T	E517X
17	1592del	I531KfsX6
Intron 17	1595+1G>A	Splice site mutation
Intron 17	1596-2A>C	Splice site mutation
Intron 17	1596-1G>C	Splice site mutation
18	1616C>G	S539X
20	2029_2031delins27	H677delinsIYX
Intron 21	2170+1G>A	Splice site mutation
Intron 22	2288+2T>C	Splice site mutation
23	EX23del	(unknown)
25	2593C>T	R865X
27	2833_2834del	K945EfsX10
29	3109A>T	K1037X
30	3157C>T	Q1053X
31	3283G>C	A1095P
33	3557_3558insAC	V1187LfsX11
34	3847del	L1283WfsX6

Exon	DNA Change ¹	Protein Change ¹
34	3889C>T	R1297X
36	4216del	V1406CfsX19
Intron 36	4242+1G>T	Splice site mutation
37	4346del	S1449FfsX4
37	4354T>C	S1452P
37	4355C>G	S1452X
37	4411C>T	R1471X
38	4419_4420insA	G1474RfsX6
39	4835del	P1612QfsX29
Intron 40	4956+1G>T	Splice site mutation
41	5253_5266del	F1751LfsX13
45	5909_5910del	E1970VfsX3
45	5920G>T	E1974X
46-50	EX46_EX50del	(unknown)
46	6059del	P2020LfsX8
46	6094C>T	R2032X
47	6283del	S2095QfsX9
48	6404_6405insT ³	S2136KfsX1
48	6419C>G	S2140X
48	6494G>A	W2165X
48	6700C>T	R2234X
Intron 48	6775-2A>C	Splice site mutation
49	6804_6805insG	S2269VfsX6
49	6828del	V2277LfsX11
50	7005G>A	W2335X
53	7339_7340insT	Y2447LfsX4
53	7378T>C	W2460R
55	7806G>A	Splice site mutation
56	7867C>T	R2623X

Exon	DNA Change ¹	Protein Change ¹
57	7985_7989del	P2662RfsX5
57	8007del	K2669NfsX21
57	8016G>C	K2672N
59	8162A>G	Y2721C
60-61	EX60_EX61del ⁴	V2738_K2824delinsAfsX4
61	8390del	G2797DfsX1
Intron 61	8472-1G>C	Splice site mutation
Intron 65	8907+2T>A	Splice site mutation
68	9109C>T	R3037X
70-73	EX70_EX73del ⁵	(unknown)
70	9190del	V3064SfsX16
70	9219C>G	Y3073X
Intron 70	9275+1G>A	Splice site mutation
Intron 70	9276-2A>T	Splice site mutation
71	9289_9290insTTTG	T3098CfsX11
71	9367del	V3123FfsX13
Intron 71	9399+2_+8del	Splice site mutation
72	9429_9432del	R3143SfsX4
72	9431_9432del	E3144VfsX5
72	9474G>A	Splice site mutation

1. Nucleotides and amino acids are numbered according to the cDNA sequence of *VPS13A/CHAC* isoform A reported by Rampoldi et al (2001), Genbank accession no. NM_033305. Mutation nomenclature is as recommended by the Human Genome Variation Society.
2. OMIM allelic variant 605978.001
3. OMIM allelic variant 605978.002
4. OMIM allelic variant 605978.003
5. Deletion extends into the neighboring gene, removing exons 6 & 7 of *GNA14* [Dobson-Stone et al 2005]